Scholars Journal of Medical Case Reports
Abbreviated Key Title: Sch J Med Case Rep
ISSN 2347-9507 (Print) | ISSN 2347-6559 (Online)
Journal homepage: https://saspublishers.com

OPEN ACCESS

Internal Medicine

Ebstein Anomaly in the Adult Patient: Case Report and Review of the Literature

Fatimazahrae Kaddari[1*], Meriam Yahyaoui[1], Mouna Zahlane[1], Laila Benjillali[1], Lamiaa Essadouni[1], Othmane Benlafkih[2], Saloua Elkarimi[2], Mustapha El Hattaoui[2]

[1]Internal Medicine Department, Errazi Hospital, University Hospital Mohammed the VIth, Marrakesh, Morocco
[2]Department of Cardiology, University Hospital Center Mohammed VI of Marrakech, Morocco

DOI: 10.36347/sjmcr.2022.v10i06.015 | **Received:** 22.04.2022 | **Accepted:** 01.06.2022 | **Published:** 12.06.2022

***Corresponding author:** Fatimazahrae Kaddari
Internal Medicine Department, Errazi Hospital, University Hospital Mohammed the VI[th], Marrakesh, Morocco

Abstract **Case Report**

Ebstein's disease is a rare congenital malformation, its prevalence is unknown, anatomically it is characterised by low insertion of one or more leaflets of the tricuspid valve on the right ventricular, reduced size of the right ventricle and tricuspid regurgitation. The diagnosis is made by non-invasive cardiac imaging with transthoracic echocardiography. We Case report a 45-year-old woman followed for pulmonary arterial hypertension and recurrent episodes of heart failure exacerbations. She was admitted to the internal medicine department for investigation of portal thrombosis. After a cardiac re-evaluation, the discovery of severe Ebstein's disease at the surgical stage. The proposed therapeutic decision is a Glenn. The evolution is marked by a rapid clinical worsening of the patient before surgery requiring a transfer in cardiac intensive care unit and then to her death. The aim of this article is to review the manifestations of Ebstein anomaly and to highlight its Mimics. Ebstein's disease is a pathology under-diagnosed or diagnosed late because it is rare and because of the clinical presentation which is similar to other pathologies, hence the need to know how to evoke it in order to make the diagnosis.

Keywords: Ebstein Anomaly- clinical and echocardiographic Manifestations- differential diagnosis.

INTRODUCTION

Ebstein's disease is a rare malformation of the tricuspid valve, it accounts for 1% of all heart defects [1], characterized by abnormal insertion of one or two leaflets of the triscupid valve. It is generally the septal and/or the posterior leaflets that are affected by this malposition while the anterior leaflet is correctly positioned. This results in a displacement of the functional orifice of the tricuspid towards the right ventricular apex or the pulmonary infundibulum, with artrialization of the right ventricle and reduction of its volume. Tricuspid leakage is observed in almost all cases and rare cases of tricuspid stenosis have been described [2].

CASE REPORT

A 45-year-old woman followed for pulmonary arterial hypertension with recurrent episodes of heart failure exacerbations. She was admitted to the internal medicine department for investigation of portal thrombosis. On admission, the patient reported NYHA stage IV dyspnea, palpitations without orthopnea or chest pain. The clinical examination showed a conscious patient hypotensive at 90/60 mmg tachycardia at 100 beats per minute, desaturation at 87% at free air, peribuccal cyanosis, erythrosis of the cheekbones, heart sounds well perceived irregular, murmur of tricuspid insufficiency, hepatomegaly, edema of the lower limbs, right basithoracic fluid effusion syndrome.

Chest X-ray showed cardiomegaly pulmonary hypo vascularization the pulmonary vascularization decreased and minimal right pleural effusion. The ECG showed atrial fibrillation.

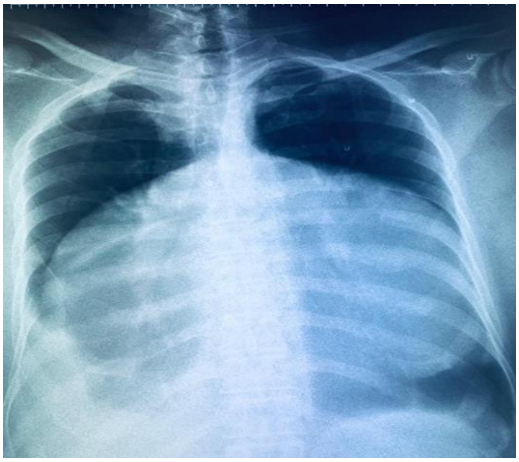

Figure 1: Chest X-ray with Cardiothoracic Index at 0.86 cm

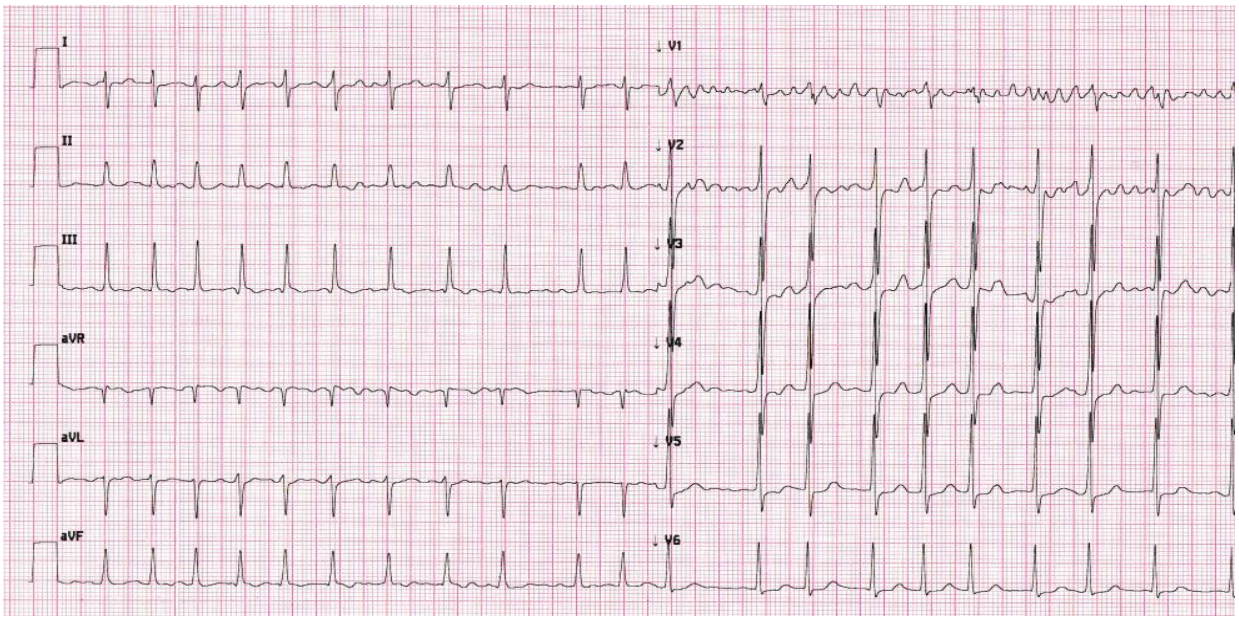

Figure 2: EKG of the patient with a mean ventricular frequency at 110

TEEs showed an aspect in favor of Ebstein's disease, rudimentary right ventricle (RV) by atrialization of its large part; ectasic right atriaum free of echo. The tricuspid valve low inserted seat of a severe tricuspid insufficiency; non dilated left ventricle (LV) and non-hypertrophied with preserved systolic function (LVEF =56 %), no significant mitroaortic valve disease, normal caliber of pulmonary artery, small pericardial effusion.

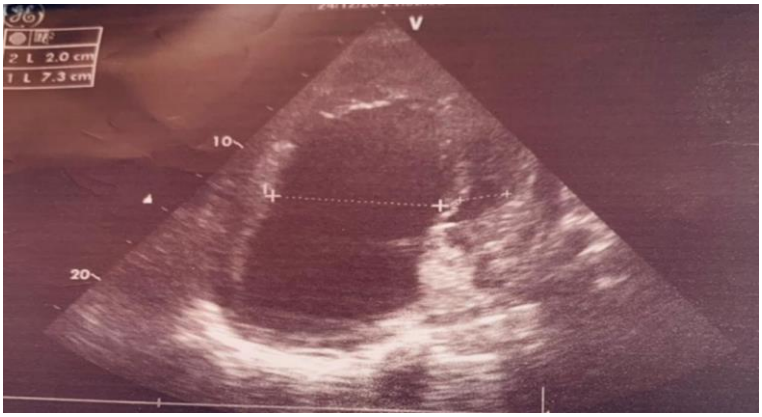

Figure 3: Cross-section of the four chambers showing atrialization of the RV with a rudimentary RV due to low tricuspid valve implantation and LV compression

Abdominal angioscanner shows a portal trunk with a small opacified appearance permeable without detectable obstruction and peritoneal effusion with moderate abondance.

The diagnosis of Ebstein's disease was retained with an indication for Glenn surgery. The evolution is marked by the aggravation of the symptomatology by signs of global cardiac insufficiency; attacks of palpitations and hypotensions requiring a transfer of the patient in intensive care unit with indication of vasoactive drug then towards her death.

DISCUSSION

The clinical presentation of Ebstein disease is heterogeneous depending on the severity of the lesion (extent of displacement of the tricuspid leaflets and their abutment) and the degree of right ventricular dysfunction. In the minor forms, patients may remain asymptomatic or have a systolic murmur, exertional dyspnea, asthenia or palpitations. In the severe forms, patients may have manifestations such as rhythm disorders, cyanosis and in some cases right heart failure. The most severe forms are found in newborns. The cardiomegaly can be such that the heart occupies the entire thorax.

In Ebstein's disease, the cardiac silhouette on the thoracic radiography may vary from normal to cardiomegaly depending on the severity of the disease.

A cardiothoracic index > 0.65cm is a predictor of a bad prognosis of the disease [3]. Our patient had clinical manifestations that may be suggestive of Ebstein's heart disease in its severe form. She had stage IV of NYHA dyspnea, cyanosis, atrial fibrillation and extrasystoles, right heart failure, systolic murmur of tricuspid insufficiency, Cardiomegaly with à cardiothoracic index at 0.86 cm.

In Ebstein disease, the diagnosis is based on echo-Doppler or three - dimensional echocardiography, which shows the bundling of the leaflets, the size of the residual functional right ventricle, as well as the site and the degree of tricuspid regurgitation and the feasibility of valve repair are also assessed by echocardiography. The diagnosis is made by measuring the apical displacement of the septal and posterior tricuspid valve leaflets relative to the anterior mitral valve. A displacement of 8 mm or more per body surface area makes the diagnosis [3].

There are several cardiopathies associated with tricuspid insufficiency and dilatation of the right cavities that may pose a differential diagnosis with Epstein's disease [4]. The most common congenital etiology of tricuspid regurgitation associated with dilatation of the right cavities in Ebstein's disease followed by tricuspid valve dysplasia [5] and Right ventricular myopathy, in particular right ventricular muscle dysplasia, which can cause tricuspid insufficiency. On the other side, rheumatic damage of the tricuspid valve [6, 7] and tricuspid valve prolapse [7, 8] are the acquired abnormalities that can lead to tricuspid regurgitation and may mimic Ebstein's disease. Less common causes of acquired anomalies and secondary regurgitation include traumatic causes [6, 7, 9], endocarditis [6, 8] myocardial infarction, carcinoid disease of the heart [7, 10, 11], radiation therapy [7, 12] and connective tissue disease [10].

The good clinical and echocardiographic evaluation makes it possible to make the difference between these various pathologies.

Ammash *et al.*, in their work 'mimics of Ebstein anomaly'. They tried to find the clinical and echographic elements that allow to distinguish the tricuspid insufficiency during the Ebstein anomaly from the other etiologies of the tricuspid regurgitation presented in the following table:

Table II. Clinical and echocardiographic clues for common non-Ebstein's TV abnormalities

TV abnormalities	Clinical clues	Echocardiographic features
Rheumatic valvular disease	Rheumatic fever, MV involvement	Focal chordal thickening, diffuse fibrous thickening, diffuse marginal or leaflet thickening of MV/TV, commissural fusions
TV prolapse	MV prolapse, straight back syndrome	Prolapse/myxomatous changes of TV/MV, TV annular dilatation
TV endocarditis	Pneumonia, intravenous drugs, habitual alcoholism, immune deficiency state	TV vegetations, ruptured chordae, valvular indentation
Traumatic tricuspid regurgitation	Chest trauma	Ruptured TV tensor apparatus (chordae, papillary muscle)
RV dysplasia	Ventricular tachycardia	Small RV aneurysms, RV trabeculations, RV dysfunction
Tricuspid annular dilation	RV infarction, primary or secondary pulmonary hypertension, cardiomyopathy	TV annular dilatation, RV enlargement and dysfunction
TV dysplasia	Remote history of heart murmur, pulmonary stenosis	Thickened-rolled leaflets, hypoplastic papillary muscle, shortened chordae
Carcinoid heart disease	Gastrointestinal hypermotility, bronchospasm, pulmonary valve involvement	Thickened margins, chordae, retracted leaflet, chordae, carcinoid plaques on ventricular aspect of TV
Connective tissue disease, radiation therapy, ergotism	Suggestive history and examination	Leaflet contraction, tethering, shortened chordae, aortic, MV involvement

MV, Mitral valve.

Figure 4: Table illustrating the clinical and echographic aspects according to the different etiologies of tricuspid insufficiency [4]

Ebstein's anomaly is a congenital cardiac malformation that can pose serious management, hemodynamic difficulties as well as electro physiologic ones.

The formal surgical indications in Ebstein's disease are clinically significant cyanosis, dyspnea stage III or IV of NYHA, heart failure, a cardiothoracic index >0.65cm on chest radiography, arrhythmia on ECG, onset of left ventricle function alteration and significant tricuspid insufficiency on transthoracic echography [1].

The case of our patient, she had a severe form of Ebstein's disease indicating an immediate surgery in front of the global cardiac decompensation requiring depletion by diuretics contrasting with severe hypotension and rhythm disorders with indication of vasoactive drugs and antiarrhythmic drugs requiring transfer to a cardiac intensive care unit while waiting for surgery then the patient died before.

CONCLUSION

Ebstein's anomaly is a rare diagnosis but should be considered as a differential diagnosis even in elderly patients with severe tricuspid regurgitation and prédominant right heart failure, in the absence of any other explanation.

RÉFÉRENCES

1. Redaction, L. (2012). Ebstein's disease: news [Internet]. Cardinal Cardiology. [cited 2021 Jan 23]. Available at: https://cardinale.fr/la- Maladie-debstein/
2. MacLellan-Tobert, S. G., Driscoll, D. J., Mottram, C. D., Mahoney, D. W., Wollan, P. C., & Danielson, G. K. (1997). Exercise tolerance in patients with Ebstein's anomaly. *Journal of the American College of Cardiology*, *29*(7), 1615-1622.
3. Celermajer, D. S., Bull, C., Till, J. A., Cullen, S., Vassillikos, V. P., Sullivan, I. D., ... & Deanfield, J. E. (1994). Ebstein's anomaly: presentation and outcome from fetus to adult. *Journal of the American College of Cardiology*, *23*(1), 170-176.
4. Ammash, N. M., Warnes, C. A., Connolly, H. M., Danielson, G. K., & Seward, J. B. (1997). Mimics of Ebstein's anomaly. *American Heart Journal*, *134*(3), 508-513.
5. Hornberger, L. K., Sahn, D. J., Kleinman, C. S., Copel, J. A., & Reed, K. L. (1991). Tricuspid valve disease with significant tricuspid insufficiency in the fetus: diagnosis and outcome. *Journal of the American College of Cardiology*, *17*(1), 167-173.
6. Waller, B. F. (1987). Etiology of pure tricuspid regurgitation. *Cardiovascular Clinics*, *17*(2), 53-95.
7. Waller, B. E., Howard, J., & Fess, S. (1995). Pathology of tricuspid valve stenosis and pure tricuspid regurgitation—part II. *Clinical cardiology*, *18*(3), 167-174.
8. Weinreich, D. J., Burke, J. F., Bharati, S., & Lev, M. (1985). Isolated prolapse of the tricuspid valve. *Journal of the American College of Cardiology*, *6*(2), 475-481.
9. Dimitrakakis, G., & Dimitrakaki, I. A. (2012). eComment. Tricuspid valve and blunt chest trauma. *Interactive cardiovascular and thoracic surgery*, *15*(3), 556-557.
10. Connolly, H. M., Nishimura, R. A., Smith, H. C., Pellikka, P. A., Mullany, C. J., & Kvols, L. K. (1995). Outcome of cardiac surgery for carcinoid heart disease. *Journal of the American College of Cardiology*, *25*(2), 410-416.
11. Hauck, A. J., Freeman, D. P., Ackermann, D. M., Danielson, G. K., & Edwards, W. D. (1988, September). Surgical pathology of the tricuspid valve: a study of 363 cases spanning 25 years. In *Mayo Clinic Proceedings* (Vol. 63, No. 9, pp. 851-863). Elsevier.
12. Brosius III, F. C., Waller, B. F., & Roberts, W. C. (1981). Radiation heart disease: analysis of 16 young (aged 15 to 33 years) necropsy patients who received over 3,500 rads to the heart. *The American journal of medicine*, *70*(3), 519-530.